**Instructions**

**September 16, 2016**

**This document contains five problems, each consisting of a brief excerpt from a genetics research article, some extra background information, and two multiple choice questions. The multiple choice questions ask you (1) to identify the argument scheme of an argument for a certain claim based upon the excerpt, and (2) to identify all and only the premises of the argument. We suggest that you look over the accompanying catalog of argument schemes first. However, you may refer to the catalog of argument schemes at any time. Do not consult external sources of information about genetics. (Our goal is for non-biologists to be able to use the catalog.) Please mark your answers on this document and return it by email to Dr. Nancy Green <**[**nlgreen@uncg.edu**](mailto:nlgreen@uncg.edu)**>.**

**Please do not share these problems or the accompanying catalog of argument schemes with anyone else. Thank you very much for participating in this study!**

1. Excerpt:

“Nephrogenic diabetes insipidus (NDI) is a [human] disease characterized by excessive urination and thirst, despite normal production of the antidiuretic hormone arginine vasopressin (AVP) [1]. The inherited forms are either X-linked as a consequence of mutation of the Avpr2 gene [2], or autosomal due to mutation of the Aqp2 gene [3] ... we found a family of mice that urinated and drank excessively ... this was an example of diabetes insipidus.”

Extra background information:

* “Autosomal” inheritance of a mutation means that the mutation was inherited from one of the first 22 pairs of chromosomes.
* “X-linked” inheritance of a mutation means that the mutation was inherited from the X chromosome of the 23rd pair of chromosomes.
* The human Avpr2 and Aqp2 genes are similar in function to the mouse Avpr2 and Aqp2 genes.

Using information from the excerpt and from the extra background information, what type of argument could you give for the following claim: ***Mutation of the Avpr2 gene or Aqp2 gene may be the cause of diabetes insipidus in this family of mice.***

Circle the argument scheme:

* Agreement
* Failed Method of Agreement
* Analogy
* Consistent Explanation
* Consistent with Predicted Effect
* Difference
* Failed Method of Difference
* Effect to Cause
* Eliminate Candidates

Circle all and only the argument’s premises:

* Humans with NDI have normal production of AVP, an antidiuretic hormone.
* Inherited forms of NDI in humans are X-linked or autosomal.
* Inherited forms of NDI in humans are caused by mutation of the Avpr2 gene or Aqp2 gene.
* The family of mice had diabetes insipidus, which is a disorder similar to nephrogenic diabetes insipidus in humans
* The human Avpr2 and Aqp2 genes are similar in function to the mouse Avpr2 and Aqp2 genes.

2. Excerpt:

“Our initial observations suggested the affected mice suffered from an apparently paroxysmal movement disorder ... At initial examination, a human movement disorder specialist ... likened the disorder to episodic intermittent ataxia ... Sequencing of all exons and intron–exon boundaries of Itpr1 [gene] in affected mice from the current study revealed a single mutation within Itpr1: a novel in-frame deletion of 18 bp within exon 36 (Itpr1Δ18/Δ18).”

Extra background information:

* The phrase “Itpr1Δ18/Δ18” refers to the Itpr1 gene mutation found in the affected mice.
* Exons include genetic sequences that code for proteins; introns do not.
* A deletion is a type of mutation in which part of a DNA sequence is lost.

Using information from the excerpt and from the extra background information, what type of argument could you give for the following claim: ***The Itpr1Δ18/Δ18 mutation may be the cause of the affected mice’s movement disorder.***

Circle the argument scheme:

* Agreement
* Failed Method of Agreement
* Analogy
* Consistent Explanation
* Consistent with Predicted Effect
* Difference
* Failed Method of Difference
* Effect to Cause
* Eliminate Candidates

Circle all and only the argument’s premises:

* The affected mice suffered from a movement disorder.
* The movement disorder of the mice was likened to episodic intermittent ataxia in humans.
* All exons and intron-exon boundaries of Itpr1 were sequenced.
* The affected mice were found to have a single mutation within Itpr1 (Itpr1Δ18/Δ18).
* A deletion is a type of mutation in which part of a DNA sequence is lost.

3. Excerpt:

“Aqp2F204V/F204V mice have dramatically increased urine production, in some cases producing an amount of urine in 24 h that exceeds their body weight, compared to their heterozygous or wild-type littermates. Such loss of water would rapidly lead to dehydration were it not compensated by increased water intake. Indeed, mutant [Aqp2F204V/F204V] mice also dramatically increase their water intake (Figure 1C) compared to their heterozygous or wild-type littermates.”

Extra background information:

* The phrase “Aqp2F204V/F204V mice” means “mice with two copies of the F204V mutation of the Aqp2 gene”.
* The “heterozygous littermates” of the Aqp2F204V/F204V mice had only one copy of the F204V mutation of the Aqp2 gene.
* The “wild-type littermates” of the Aqp2F204V/F204V mice had no copies of the F204V mutation of the Aqp2 gene.

Using information from the excerpt and from the extra background information, what type of argument could you give for the following claim: ***The two copies of the F204V mutation of the Aqp2 gene may be the cause of the increased urine production in the Aqp2F204V/F204V mice.***

Circle the argument scheme:

* Agreement
* Failed Method of Agreement
* Analogy
* Consistent Explanation
* Consistent with Predicted Effect
* Difference
* Failed Method of Difference
* Effect to Cause
* Eliminate Candidates

Circle all and only the argument’s premises:

* Aqp2F204V/F204V mice have two copies of the F204V mutation of the Aqp2 gene.
* Aqp2F204V/F204V mice have a disorder similar to diabetes insipidus in humans.
* Aqp2F204V/F204V mice have dramatically increased urine production compared to their heterozygous or wild-type littermates.
* The heterozygous or wild-type littermates do not have two copies of the F204V mutation of the Aqp2 gene.
* Increased urine production would lead to dehydration, unless compensated by increased water intake.

**4.** Excerpt:

“The disorder in these mice segregated in a monogenic, autosomal recessive manner, making Aqp2 a candidate gene. Sequencing of Aqp2 coding region of affected mice identified a thymine to guanine (T to G) transversion (Figure 1A), which is predicted to lead to a valine for phenylalanine substitution at amino acid 204 of the [AQP2F04] protein ... [AQP2 protein is] conserved among vertebrate species.”

Extra background information:

* The “thymine to guanine (T to G) transversion” describes the mutation in the Aqp2 gene found in mice with the disorder.
* The Aqp2 gene mutation that was found in the mice is predicted to produce AQP2F204 protein, an abnormal form of AQP2 protein.
* Conserved sequences are similar or identical sequences in DNA or protein in different species.
* Amino acid changes in conserved sequences may severely impact protein structure and function resulting in an abnormal phenotype.

Using information from the excerpt and from the extra background information, what type of argument could you give for the following claim: ***There is a plausible explanation at the molecular level of how the Aqp2 mutation may be the cause of the disorder in the affected mice.***

Circle the argument scheme:

* Agreement
* Failed Method of Agreement
* Analogy
* Consistent Explanation
* Consistent with Predicted Effect
* Difference
* Failed Method of Difference
* Effect to Cause
* Eliminate Candidates

Circle all and only the argument’s premises:

* The affected mice have a mutation in Aqp2.
* The Aqp2 mutation is predicted to produce abnormal (AQP2F204) protein.
* Conserved sequences are similar or identical sequences in DNA or protein in different species.
* AQP2 protein is conserved among vertebrate species.
* Amino acid changes in conserved sequences may severely impact protein structure and function resulting in an abnormal phenotype.

5. Excerpt (same as in #4):

“The disorder in these mice segregated in a monogenic, autosomal recessive manner, making Aqp2 a candidate gene. Sequencing of Aqp2 coding region of affected mice identified a thymine to guanine (T to G) transversion (Figure 1A), which is predicted to lead to a valine for phenylalanine substitution at amino acid 204 of the [AQP2F04] protein ... [AQP2 protein is] ... conserved among vertebrate species.”

Extra background information:

* The affected mice have a disorder similar to nephrogenic diabetes insipidus (NDI) in humans.
* The “thymine to guanine (T to G) transversion” describes the mutation in the Aqp2 gene found in mice with the disorder
* The Aqp2 mutation that was found in the mice is predicted to produce AQP2F204 protein, an abnormal form of AQP2 protein.
* The mouse Aqp2 gene is functionally similar to the human AQP2 gene.
* Amino acid changes in conserved sequences may severely impact protein structure and function resulting in an abnormal phenotype.

Using information from the excerpt and from the extra background information, what type of argument could you give for the following claim: ***There is a plausible explanation at the molecular level of how AQP2 mutation may cause nephrogenic diabetes insipidus (NDI) in humans.***

Circle the argument scheme:

* Agreement
* Failed Method of Agreement
* Analogy
* Consistent Explanation
* Consistent with Predicted Effect
* Difference
* Failed Method of Difference
* Effect to Cause
* Eliminate Candidates

Circle all and only the argument’s premises:

* The affected mice have a disorder similar to nephrogenic diabetes insipidus (NDI) in humans.
* The Aqp2 mutation in the affected mice is predicted to produce abnormal (AQP2F204) protein.
* The mouse Aqp2 gene is functionally similar to the human AQP2 gene.
* AQP2 protein is conserved among vertebrate species.
* Amino acid changes in conserved sequences may severely impact protein structure and function resulting in an abnormal phenotype.